

Table 4. Summary of Sequence Variations in *SIX3*

	Sequence Change	Expected Effect	Type of Mutation	Reference
Mutations	676C→G	L226V	Missense	Wallis et al 1999
	696-705 del	232-234 del	Deletion in frame	Wallis et al 1999
	749T→C	V250A	Missense	Wallis et al 1999
	770C→G	R257P	Missense	Wallis et al 1999
Polymorphisms	Sequence Change	Expected Effect	Frequency	Reference
	90G→T	A30A	3/326	Present study
	219C→T	P73P	1/326	Present study
	576C→T	R192R	27/351 ¹	Nanni et al 2000
	942G→A	A314A	34/315 ²	Present study

1. 2 individuals with 576C→T are homozygous T.

2. 1 individual with 942G→A is homozygous A; 15 individuals are heterozygous for both 576C→T and 942G→A.

References

Nanni L, Croen LA, Lammer EJ, Muenke M (2000) Holoprosencephaly: molecular study of a California population. *Am J Med Genet* 90:315-9 [[Medline](#)]

Wallis DE, Roessler E, Hehr U, Nanni L, Wiltshire T, Richieri-Costa A, Gillessen-Kaesbach G, Zackai EH, Rommens J, Muenke M (1999) Mutations in the homeodomain of the human *SIX3* gene cause holoprosencephaly. *Nat Genet* 22:196-8 [[Medline](#)]